Note: The following information is provided by the author(s) and has not been reviewed by GeneReviews staff.

Table 4. Pathologic Allelic Variants in *RYR1* Associated with Multiminicore Disease

Nucleotide Change	Amino Acid Change	Exon
c.212A>C	S71Y	3
c.325C>T	R109W	4
c.1453A>G	M485V	14
c.4729G>A	A1577T	33
c.6178G>T	G2060C	38
c.6847A>C	N2283H	42
c.7268T>A	M2423K	45
c.8816G>A	R2939K	57
c.10343C>T	S3448F	68
c.10579C>T	P3527S	71
c.11315G>A	R3772Q	79
c.12986C>A	A4329D	91
c.14126C>T	T4709M	96
c.14365-2A>T	Acceptor splice site mutation	Intron 99
c.14545G>A	V4849I	101
14646+2.99 kb A>G (splice site mutation)	Frameshift additional exon of 94 AA; premature stop codon 4976X	101-102